Cutaneous hyperpigmentation and cobalamin deficiency

An exclusively breastfed five-month-old female was referred with anaemia, pallor and hyperpigmented genitalia and inner thighs (top). She was also lethargic, irritable and showed failure to thrive. Laboratory studies revealed a macrocytic anaemia with haemoglobin concentration (Hb) 73 g/l (normal range 95–140), mean corpuscular volume (MCV) 104 fl (85–97) and neutropenia $0{\cdot}52 \times 10^9$/l (1·0–5·6). The blood film showed macrocytosis, rare poikilocytes and neutropenia. Serum vitamin B_{12} was undetectable, with high plasma homocysteine [78·8 μmol/l (3·7–13·9 μmol/l)]) and negative anti-parietal cell and anti-intrinsic factor antibodies. Her mother's laboratory evaluation showed Hb 132 g/l (115–150), MCV 93·6 fl (80–97) and undetectable serum vitamin B_{12}, with antibodies to parietal cells and intrinsic factor being detected. Intramuscular* hydroxocobalamin was started, with progressive haematological and clinical improvement. The infant's vitamin B_{12} level was normal on follow-up evaluation 2 months after diagnosis and the hyperpigmentation gradually improved (bottom).

Early diagnosis of neonatal B_{12} deficiency is very important as infants can develop severe neurological sequelae. Mothers are frequently asymptomatic and nonspecific clinical findings are easily overlooked. Cobalamin deficiency affects all age groups and multiple systems, namely the bone marrow and the gastrointestinal and nervous systems. Mucocutaneous signs are less common but can predate other manifestations. In fact, this deficiency must be considered in the differential diagnosis of cutaneous hyperpigmentation. Treatment usually reverses the abnormalities, but some may persist.

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